Don't put anything smaller than your elbow in your ear

the genetics of ear wax

Created: October 11, 2006

Recently, an exciting genetic discovery was made in the field of ear wax. It appears that a change in a single nucleotide of your DNA can determine whether your ear wax is wet or dry. This marks the first time that a single-nucleotide polymorphism (SNP) has been found to determine a visible genetic trait.

Known medically as cerumen, ear wax is found in the ears of humans and many other mammals. Secreted by apocrine glands in the ear canal, the exact function of ear wax is not clear. Proposed benefits range from keeping the ear canal clean and lubricated, to acting as an insect trap.

In contrast, many have experienced the disadvantages of ear wax. In those who are hard of hearing, ear wax can damage their hearing aids. Ear wax is also a cause of hearing loss—a build up of wax can block the ear canal, hindering the passage of sound waves to the ear drum.

But it has long been noted that there are 2 types of ear wax, "wet" and "dry". Wet ear wax common in Caucasians and African-Americans, it tends to be honey-to-brown in color and sticky in nature. In contrast, dry ear wax is common in East Asians and is gray in color and more brittle and flakey. And now we know the underlying genetic basis.

A major clue came in 2002 when scientists reported the case of a Japanese woman who had a rare genetic disorder that caused her arms and legs to twist uncontrollably (paroxysmal kinesigenic choreoathetosis). Uncommon in the Japanese, she also happened to have wet ear wax, as did several of her relatives who also had choreoathetosis. This suggested that the inheritance of the rare neurological disorder and the inheritance of the dry ear wax type were linked.

Linkage anaylsis of these 2 traits in affected family members pointed to the pericentromeric region of chromosome 16 as the site that contained both the "choreoathetosis locus" and the "ear wax locus". To isolate the specific genetic change that determined ear wax type, scientists compared the sequence of this region in Japanese people with dry ear wax with the sequence of Japanese people with wet ear wax. They found 3 SNPs which were associated with ear wax type, spread over a 5-gene region of DNA.

But only 1 of the SNPs, found in the ABCC11 gene (Bookshelf, PubMed Central, PubMed, OMIM, Entrez Gene), resulted in a nonsynonymous change in the protein product. The SNP, found in the ABCC11 gene, is a G538A substitution (in the reference sequence rs17822931) and results in a G180R substitution in the transporter protein it encodes. Individuals who inherit at least 1 copy of guanine at position 538 (GG homozygotes and GA heterozygotes) have wet ear wax, a dominant trait. Individuals with dry ear wax, a recessive trait, are AA homozygotes.

It is possible that the ABCC11 protein transports some of the lipids and granules found in wet ear wax, and a change of just one of its amino acids results in the production of dry ear wax that lacks some of these molecules.

Around the world, the "dry" A allele is more common in Asia, being most common in the North and East of Asia (100% in Northern Han Chinese and Koreans, less high in Mongolians, other areas of China, and Japan). Having apocrine glands that secrete dry ear wax may also be linked to decreased sweating from apocrine glands under the arms, and a decrease in bodily secretions in colder climates could be a survival advantage.

The A allele is also found in Native Americans, possibly reflecting the migration of their Ancestors from Siberia to North America.

In contrast, the "wet" G allele is more common in Sub-Saharan Africans and Caucasians.

Regardless of whether the ear wax is wet or dry, the ear has the same built-in conveyer-belt mechanism for ridding itself of cerumen—it uses epithelial cells that migrate away from the ear drum and up the ear canal to transport ear wax, and any debris it contains, out of the ear. These cells migrate at speeds of about 0.05 mm/day (similar to the rate of nail growth).

Many problems with ear wax result from people using cotton tips in an attempt to remove the wax. But what they actually do is push the wax further into the ears, against the tide of migrating cells, causing it to impact and accumulate. So, to quote the words said by many Ear, Nose, and Throat (ENT) surgeons, "Do not put anything smaller than your elbow in your ear!".

Let nature take its course, or see a doctor instead.

References

- Yoshiura K, Kinoshita A, Ishida T, Ninokata A, Ishikawa T, Kaname T, Bannai M, Tokunaga K, Sonoda S, Komaki R, et al. A SNP in the ABCC11 gene is the determinant of human earwax type. Nature Genetics 38(3):324–330; 2006. (PubMed)
- Tomita H, Yamada K, Ghadami M, Ogura T, Yanai Y, Nakatomi K, Sadamatsu M, Masui A, Kato N, Niikawa N. Mapping of the wet/dry earwax locus to the pericentromeric region of chromosome 16. Lancet 359(9322):2000–2002; 2002. (PubMed)

Link Roundup

NCBI Literature Resources

In OMIM, read about the ABCC11 gene and the ear wax phenotype

Search for more information about ABCC11 in: - Bookshelf - PubMed Central - PubMed

Additional NCBI Resources

Where in the genome does ABCC11 lie? MapViewer

Are similar sequences found in other species? Homologene

Where can I find out more information about the G538A polymorphism in ABCC11? SNP [http://www.ncbi.nlm.nih.gov/SNP/snp_ref.cgi?rs=17822931]

Where can I view the coding sequence of the ABCC11 gene? Visit Entrez Protein [http://www.ncbi.nlm.nih.gov/entrez/viewer.fcgi? db=protein&val=24660220] and click on "CDS" [http://www.ncbi.nlm.nih.gov/entrez/viewer.fcgi? val=24660219&itemID=1&view=gbwithparts]Scroll down to "ORIGIN" to see the coding sequence. At position 538, there is the wildtype guanine (G).